Appendix 5

1: Hum Mol Genet. 2003 Sep 1; 12(17): 2109-20.

Related Articles, Links

A mouse model for cystinuria type I.

Peters T, Thaete C, Wolf S, Popp A, Sedlmeier R, Grosse J, Nehls MC, Russ A, Schlueter V.

Ingenium Pharmaceuticals AG, Fraunhoferstr. 13, 82152 Martinsried, Germany.

Cystinuria, one of the most common inborn errors of metabolism in humans, accounts for 1-2% of all cases of renal lithiasis. It is caused by defects in the heterodimeric transporter system rBAT/b0,+AT, which lead to reduced reabsorption of cystine and dibasic amino acids through the epithelial cells of the renal tubules and the intestine. In an N-ethyl-N-nitrosourea mutagenesis screen for recessive mutations we identified a mutant mouse with elevated concentrations of lysine, arginine and ornithine in urine, displaying the clinical syndrome of urolithiasis and its complications. Positional cloning of the causative mutation identified a missense mutation in the solute carrier family 3 member 1 gene (Slc3a1) leading to an amino acid exchange D140G in the extracellular domain of the rBAT protein. The mouse model mimics the aetiology and clinical manifestations of human cystinuria type I, and is suitable for the study of its pathophysiology as well as the evaluation of therapeutic and metaphylactic approaches.

PMID: 12923163 [PubMed - in process]

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